



African L0a mtDNA haplogroup in Republic of Macedonia

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ABSTRACT

Mitochondrial DNA analysis from individuals from Republic of Macedonia as one of the south-eastern European countries showed that the phylogenetic characteristics of Macedonian population are similar as in other European countries, sharing west Eurasian haplogroups, predominantly European haplogroup H. Macedonian population is multiethnic and some of this ethnic groups have specific mtDNA haplogroup traced to their origin. In this article we present first finding of an African haplogroup L0a in orthodox Macedonian. From the published data it is evident that most likely L lineages arrived in Europe in rather recent historical times, what for today's Republic of Macedonia from historical point of view could be in the last 2500 years.

1. Introduction

The distribution of mtDNA haplogroups in Macedonia is similar as in other countries of Balkan Peninsula, with the most frequent European haplogroup H, than clusters U,T,J and K observed with intermediate frequency [1]. As a multiethnic country there are some haplogroups specific for ethnic groups such as M5a1 for Romanies [2] or H12 for Albanians [3]. Presence of sub-Saharan mtDNA haplogroup L0a in an orthodox Macedonian family can be explained with later arrival of sub-Saharan individuals in Europe and their dispersal throughout Europe [4,5]. The territory of today's Republic of Macedonia in the last 2500 years have been part of tumultuous historical happenings and it was part of empires that encompassed huge areas including north of Africa, Middle and Near East allowing separate arrival of south African haplogroup L0a individuals on the territory of Republic of Macedonia.

Historical periods that can be pointed out as possible for separate arrivals of individuals with sub-Saharan L0a haplogroup on the territory of today's Republic of Macedonia are following: the period of the empire of Aleksandar the Great which arose and fell 350–150 years BC which included the territory of south-east Europe, north Africa, Middle East up to India, Roman empire from 150 years BC till 400 years AD which encompassed territory of Europe and big part of Africa and Near East and 1000 years after Roman empire Eastern Roman empire when the crusading war happened and the roads toward and from Near East were passing through territory of now day Republic of Macedonia. Also should be mentioned the Ottoman Empire existing from early 14th century till 20th century encompassing southeast Europe, little Asia,

North Africa, Near East and some parts of Middle East.

2. Material and methods

2.1. Sample collection and DNA extraction

Sample was collected with buccal swab. The individual that was sampled is from central part of Macedonia, from orthodox Macedonian family. DNA was extracted using QIAamp mini kit (Qiagen, Hilden, Germany) following the manufacturer's recommendations.

2.2. Mitochondrial DNA amplification, sequencing and data analysis

Mitochondrial DNA ranges 16024–16365 and 73–340 were amplified using L15997/H16401 for HVR-I and L00029/H00408 for HVR-II primers under conditions given by Parson et al. [6]. Amplicons were purified with MiniElute PCR purification kit (Qiagen, Germany), templates were sequenced in both directions, in the forward direction with primer L15997 and in the reverse direction with primer H16401 for HVR-I, and with L00029 and H00408 for HVR-II using BigDye Terminator Cycle Sequencing Ready Reaction Kit (Applied Biosystems). Sequencing products were purified using Centriscip Spin columns (Princeton USA). Electrophoresis was carried out on Genetic Analyzer 3500 (Applied Biosystems) and analysis were performed using Sequencing software version 5.4. and sequences were aligned [7] and analyzed with SeqScape software version 2.7 (Applied Biosystems). The mtDNA haplotype was determined using EMPOP mtDNA database, v3 (www.empop.org) [8].

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Table 1
Polymorphisms within given ranges of mtDNA determining haplogroup L0a in Macedonian orthodox family.

Range 16024–16365	Range 73–340
T16086C	A93G
G16129A	C151T
C16148T	T152C
A16166G	G185A
C16168T	A189G
T16172C	A200G
C16187T	T236C
C16188G	G247A
T16189C	A263G
C16223T	309.1C
A16230G	315.1C
T16263C	
T16311C	
C16320T	

3. Results and discussion

Observed mtDNA polymorphisms from the sample determining a haplogroup L0a are given in Table 1. L0a is a clade appeared from clade L0abf after split of L0abfk clade into L0abf and L0k [9] which belong to L0 haplogroup, a daughter branch of mtDNA root. L0a may have eastern African origin. Sporadic population movements of sub-Saharan

haplogroup L0a appear to have contributed to the European mitochondrial pool. In the last few thousand years within the modern human migrations to European destinations, the territory of today's Macedonia in separate historical times could have been a place of individual settlements.

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